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LETTER TO THE EDITOR

Sexual Function

Anejaculation in a patient with Charcot–Marie–Tooth

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Dear Editor,

The occurrence of delayed ejaculation or anejaculation has been previously suggested in patients with Charcot–Marie–Tooth (CMT) syndrome.[1] Despite this, such disorder is rarely investigated and may be underestimated in patients with this disease. We report the case of a 20-year-old men with CMT 1B due to the presence of a punctiform mutation in the exon 2 of the myelin protein zero (*MPZ*) gene (1q23.3) (Ser78Leu), complaining for life-long anejaculation. He inherited this mutation from the mother and it has also been detected in his grandmother. No other components of his family were positive at the genetic testing.

This study was approved by the Intradivisional Ethics Committee of the Andrology Section, and the informed consent was provided by the patient for the publication of his clinical data. The patient was able to achieve the orgasm. At the andrological examination, the testicular volume was of 10 ml bilaterally, and the epididymis was of normal shape and consistency. The vas deferens was present bilaterally. Secondary sexual characters were normally represented. The hormonal profile (luteinizing hormone, follicle-stimulating hormone, prolactin, and total testosterone) did not reveal any abnormality. At the urine examination, performed for two different occasions after orgasm, no spermatozoa were found. The prostate-vesicular ultrasound examination showed, at baseline, a normal prostate volume; the ampulla of the ductus deferens was present and not dilated bilaterally. The seminal vesicles were not dilated; the fundus anteroposterior diameter (APD) of the right seminal vesicle was 10 mm, and 11 mm was that of the left seminal vesicle. In both seminal vesicles, the fundus APD was remeasured after orgasm: on the right seminal vesicle it was 8 mm and on the left one it was 9 mm. The difference between the fundus APD measured at baseline and after orgasm was lower than 3 mm (**Figure 1**). According to a previous study,[2] the ultrasound evaluation was compatible with a bilateral atony of seminal vesicles. The pudendal nerve somatosensory evoked potentials (SEP) showed a dysfunction occurring at the level of the peripheral nerve, while at the level of the spinal cord and the cortex, the examination did not show any abnormality. The patient was able to achieve the erection through masturbation and did not complain about erectile dysfunction (ED). Accordingly, the gland sensitivity, evaluated by penile biothesiometry, did not reveal any abnormality.

During ejaculation, the autonomic nervous system (ANS) controls the emission phase in healthy men. Postganglion noradrenergic neurons from the hypogastric nerve innervate vas deferens, seminal vesicles, and prostate and the bladder neck. Noradrenaline release increases the intraluminal pressure of such districts, carrying spermatozoa into the posterior urethra.[3] Previous authors suggested that anejaculation could be due to the psychological factors in these patients.[1] However, we showed the presence of seminal vesicles atony in a patient with CMT 1B carrying the *MPZ* Ser78Leu mutation. In addition, neurogenic bladder has been described in CMT patients with *MPZ* mutations[4,5] and a prevalent parasympathetic nervous system involvement has been observed, especially in patients with the *MPZ* Thr124Met mutation.[4] Since ejaculation occurs after noradrenaline release in postganglion synaptic space in healthy men, we speculate that *MPZ* mutations, by affecting the ANS and the balance between sympathetic and parasympathetic signals, may negatively affect seminal vesicles emptying, leading to anejaculation. Consistent with this hypothesis, infertility has never been described in CMT patients. Thus, anejaculation may selectively regard some CMT types. However, further studies are needed to timely estimate the prevalence of anejaculation in patients with CMT and its association, if any, with CMT types.

The presence of urogenital and sexual dysfunction has been already shown in women with CMT.[5,6] Lower urinary tract dysfunction (LUTS),[7]

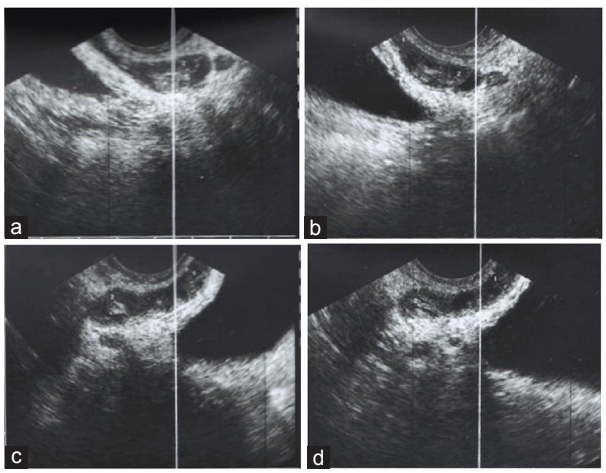

Figure 1: Seminal vesicles' measures at baseline and after orgasm. Right seminal vesicle at baseline (**a**) and after orgasm (**b**). At baseline, the anteroposterior diameter in the fundus was 10 mm and in the cauda it was 5 mm. After orgasm, these measures were 8 mm and 4 mm, respectively. Left seminal vesicle at baseline (**c**) and after orgasm (**d**). At baseline, the anteroposterior diameter in the fundus was 11 mm and in the cauda it was 6 mm; after orgasm, these measures were 9 mm and 6 mm, respectively.

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neurogenic bladder, ED, and sexual dysfunctions have been reported in male patients with CMT,[6–8] but the prevalence of such disturbances in patients with CMT is unknown. Unfortunately, the literature lacks of studies describing ejaculatory disorders in these patients. Furthermore, it should be kept in mind that the major of studies used questionnaires to evaluate the presence of urogenital dysfunction,[6,7] resulting in contradictory data. The presence of ED and anejaculation needs to be investigated through more objective methods, such as penile Doppler evaluation, basal and postorgasm prostate vesicular ultrasound examination, and penile biothesiometry. This would help to better understand the prevalence of urogenital dysfunctions in men with CMT.

In conclusion, we reported the presence of anejaculation, atony of seminal vesicles, and a dysfunction occurring at the level of the peripheral nerve in a patient with CMT 1B. The penile sensitivity was not affected. Most of the studies have not investigated this topic, probably due to the difficulty for both physicians and patients to discuss about sexuality. It may also be hypothesized that such problems might not be considered of priority in patients with CMT.[1,7] However, urogenital dysfunction heavily affects the quality of life, and both LUTS and ED have already been detected in patients with CMT.[7,8] Therefore, we believe that uro-andrologic counseling should be added in the global diagnostic workup of patients with CMT starting from the adolescence. Indeed, it may help to point out sexual disorders (such as erectile and/or ejaculation dysfunction) which might be hidden by the patient. The establishment of the proper therapeutic strategy and the consequent amelioration of the disorder might positively affect their quality of life.

AUTHOR CONTRIBUTIONS

RC and AEC designed the research study and wrote the paper, GB performed the prostate-vesicular ultrasound examination, and ESV and SLV gave a substantial contribution to the analysis and interpretation of data; all authors read and approved the final version of the manuscript.

COMPETING INTERESTS

All authors declared no competing interests.

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